

## GENETIC DISEASES EVALUATION CENTER

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### Test Name & Code:

- **Test Name:** ACTN3 Mutation Analysis
- **Test Code:** 5210

### Related Diseases & Genes:

- Alpha-actinin-3 deficiency (OMIM #: Sprinting performance) || Gene: ACTN3 (OMIM #: 102574)

### Synonyms:

- "ACTININ, ALPHA-3"
- Alpha Actinin R577X Polymorphism Analysis

### Test Details:

- **Methodology:** PCR-RFLP
- **Performed:** Everyday
- **Reported:** 7-10 days

### Specimen Details:

- **Specimen Collection:** Peripheral blood
- **Container:** Tube with EDTA
- **Specimen Volume/Amount:** > 5 ml
- **Storage/Transport Condition:** Refrigerated
- **Unacceptable Specimens/Conditions:** Serum; Frozen specimens; Hemolyzed specimens; Icterus; Lipemia

### Testing Area(s):

- Predisposition/Risk Factors

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